



## ARSE gene

arylsulfatase E (chondrodysplasia punctata 1)

### Normal Function

The *ARSE* gene provides instructions for making an enzyme called arylsulfatase E. This enzyme is part of a group known as sulfatases, which are enzymes that help process molecules that contain chemical groups known as sulfates. Sulfatases play important roles in cartilage and bone development.

Within cells, arylsulfatase E is located in the Golgi apparatus, a structure that modifies newly produced enzymes and other proteins. The function of this enzyme is unknown, although researchers believe it participates in a chemical pathway involving vitamin K. Evidence suggests that vitamin K normally plays a role in bone growth and maintenance of bone density.

### Health Conditions Related to Genetic Changes

#### X-linked chondrodysplasia punctata 1

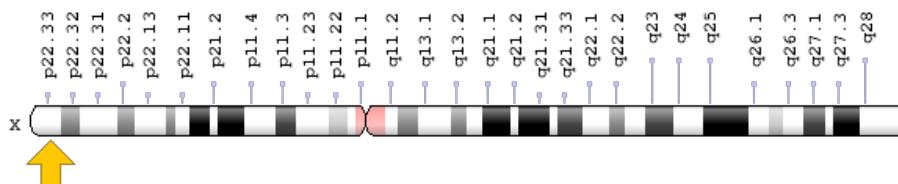
Genetic changes involving the *ARSE* gene are responsible for X-linked chondrodysplasia punctata 1, a disorder of bone and cartilage development that occurs almost exclusively in males. Between 60 and 75 percent of males with the characteristic features of this condition have a mutation within the *ARSE* gene. At least 18 mutations have been found in affected individuals; these genetic changes reduce or eliminate the function of arylsulfatase E. Another 25 percent of affected males have a small deletion of genetic material from the region of the X chromosome that contains the *ARSE* gene. These individuals are missing the entire gene, so their cells produce no functional arylsulfatase E.

It is unclear how a shortage of arylsulfatase E disrupts the development of bones and cartilage and leads to the characteristic features of X-linked chondrodysplasia punctata 1.

## Chromosomal Location

Cytogenetic Location: Xp22.33, which is the short (p) arm of the X chromosome at position 22.33

Molecular Location: base pairs 2,934,632 to 2,968,310 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ARSE\_HUMAN
- arylsulfatase E
- CDPX
- CDPX1
- CDPXR
- MGC163310

## Additional Information & Resources

### Educational Resources

- Developmental Biology (sixth edition, 2000): Osteogenesis: The Development of Bones  
<https://www.ncbi.nlm.nih.gov/books/NBK10056/>

### GeneReviews

- Chondrodysplasia Punctata 1, X-Linked  
<https://www.ncbi.nlm.nih.gov/books/NBK1544>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARSE%5BTIAB%5D%29+OR+%28arylsulfatase+E%5BTIAB%5D%29%29+OR+%28CDPX1%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- ARYLSULFATASE E  
<http://omim.org/entry/300180>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ARSE.html](http://atlasgeneticsoncology.org/Genes/GC_ARSE.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ARSE%5Bgene%5D>
- HGNC Gene Family: Sulfatases  
<http://www.genenames.org/cgi-bin/genefamilies/set/410>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=719](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=719)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/415>
- UniProt  
<http://www.uniprot.org/uniprot/P51690>

## **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/ARSE>

Reviewed: November 2011

Published: March 21, 2017

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